

Amendments to the Specification

Please replace the Table 3 on page 106 with the replacement Table 3 on the following pages:

Table 3

Mutation	Exon	Exon size	Pop Freq	Location	Sequence Around Mutation Site (SEQ. ID. Nos. 128-191)	PRIMERS	Amp Sz.
297-3 C>T	2	109	Manchester	Sub C>T + 3 Exon 3	CTTTTATTC TTTTG(C>T)AGAG AATGGGATAG A	787/788	297
R75Q	3	109	Manchester	Substitute G>A at 60	TAATGCCCTT CGGC(G>A)ATGTT TTTTGTGGA	787/788	297
300 del A	3	109	Manchester	Delete A at 4	ATTCCTTTTGC AGAGAA TGGG ATAGAGAGCT GGCT	787/788	297
E60X	3	109	Manchester	Substitute G>T at 14	GAATGGGATA GA (G>T) AGCTGGC TTCAAAGA	787/788	297
L88S	3	109	Manchester	Substitute T>C at 99	CTATGGAATC TTTT(T>C)ATATT TAGGGGTAAG	787/788	297
G86E	3	109	0.70%	Substitute G>A at 90	TTATGTTCTA TG(G>A)AATCTTT TTATATTTAG	787/788	297
R117H	4	216	0.80%	Substitute G>A at 77	AACAAGGAGG AAC(G>A)CTCTAT CGCGATTTAT	851/769	381
R117C	4	216	rare	Substitute C>T at 76	AACAAGGAGG AA(C>t)GCTCTAT CGCGATTTAT	851/769	381
Y122X	4	216	0.30%	Substitute T>A at 93	TATCGCGATT TA(T>A)CTAGGCA TAGGCTTATG	851/769	381
1148T	4	216	Fr Can (10%)	Substitute T>C at 170	GGCCTTCATC ACA(T>C)TGGAAT GCAGATGAGA	851/769	381
621+1G>T	4	216	1.30%	Sub G>T after last base	GATTTATAAG AAG(G>T)TAATAC TTCCTTGCAC	851/769	381
711+1G>T	5	90	0.90%	Sub G>T after last base	CAAAATTTGAT GAA(G>t)TATGTA CCTATTGATT	887/888	289
L206W	6a	164	Fr Can (10%)	Substitute T>G at 38	TGGATCGCTC CTT(T>G)GCAAGT GGCACTCCTC	934/935	331
1138 ins G	7	247	Manchester	Insert G at 137	AATCATCCTC CGGAAAgATA TTCACCACCA TCT	789/790	404
1154 ins TC	7	247	Manchester	Insert TC at 153	TATTCACCAC CATCTCtctAT TCTGCATTGT T	789/790	404

Mutation	Exon	Exon size	Pop Freq	Location	Sequence Around Mutation Site (SEQ. ID. Nos. 128-191)	PRIMERS	Amp Sz.
1161 del C	7	247	Manchester	Delete C at 160	CCACCATCTC ATTCTGcATT CTTCTGCGCA TGG	789/790	404
R334W	7	247	0.40%	Substitute C>T at 131	AAGGAATCAT CCTC(C>T)GGAAA ATATTTCATTA	789/790	404
R347H	7	247	0.10%	Substitute G>A at 171	CTGCATTGTT CTGC(G>A)CATGG CGGTCACTCG	789/790	404
R347L	7	247	rare	Substitute G>T at 171	CTGCATTGTT CTGC(G>T)CATGG CGGTCACTCG	789/790	404
R347P	7	247	0.50%	Substitute G>C at 171	CTGCATTGTT CTGC(G>c)CATGG CGGTCACTCG	789/790	404
1078 del T	7	247	1.10%	Delete T at 77	CTTCTTCTCA GGGTTCTTGT GGTGTTTTTA TC	789/790	404
1248 + 1 G>A	7	247	Manchester	Sub G>A1 after Exon 7	AAACAAAATA CAG(G>A)TAATGT ACCATAATG	789/790	404
A455E	9	183	0.40%	Substitute C>A at 155	AGGACAGTTG TTGG(c>a)GGTTG CTGGATCCA	891/892	386
G480C	10	192	rare	Substitute G>T at 46	GGAGCCTTGA CAG(G>T)GTAAAA TTAAGCACa	760/850	304
O493X	10	192	0.30%	Substitute C>T at 85	TCATTCTGTT CT(C>T)AGTTTTC CTGGATTAT	760/850	304
DI1507	10	192	0.50%	Delete 126, 127, 128	ATTAAAGAAA ATATcatCTT TGGTGTTTCC TATG	760/850	304
F508C	10	192	rare	Substitute T>G at 131	TAAAGAAAAT ATCATCT(T>g)TG GTGTTTCCTA	760/850	304
DF508	10	192	67.20%	Delete 129, 130, 131	ATTAAAGAAA ATATCATcTG GTGTTTCCTA TG	760/850	304
V520F	10	192	0.20%	Substitute G>T at 166	TAGATACAGA AGC(G>T)TCATCA AAGCATGCC	760/850	304
1717-1G>A	10	95	1.10%	Sub G>A at +1 Ex11	TATTTTGGT AATA(G>a)GACAT CTCCAAGTTT	762/763	233
G542X	11	95	3.40%	Substitute G>T at 40	ACAATATAGT TCTT(G>T)GAGAA GGTGGAAAT	762/762	233
S549N	11	95	rare	Substitute G>A at 62	AGGTGGAATC ACACTGA(G>A)TG GAGGTCAACG	762/763	233

Mutation	Exon	Exon size	Pop Freq	Location	Sequence Around Mutation Site (SEQ. ID. Nos. 128-191)	PRIMERS	Amp Sz.
S549I	11	95	rare	Substitute G>T at 62	AGGTGAATCA CACTGA(G>T)TGG AGGTCAACG	762/763	233
S549R (A>C)	11	95	rare	Substitute A>C at 61	AGGTGGAATC ACAC TG(A>c)GTG GAGGTCAACG	762/763	233
S549R (T>G)	11	95	0.30%	Substitute T>G at 63	AGGTGGAATC ACAC TGAG(T>G)G GAGGTCAACG	762/763	233
G551D	11	95	2.40%	Substitute G>A at 88	ATCACACTGA GTGGAG(G>A)TCA ACGAGCAAGA	762/763	233
G551S	11	95	rare	Substitute G>A at 67	ATCACACTGA GTGGA(G>A)GTCA ACGAGCAAGA	762/763	233
O552X	11	95	rare	Substitute C>T at 70	ACACTGAGTG GAGGT(C>T)AACG AGCAAGAAAT	762/763	233
R522Q	11	95	rare	Substitute G>A at 74	TGAGTGGAGG TCAAC(G>A)AGCA AGAAATTTCT	762/763	233
R563X	11	95	1.30%	Substitute C>T at 73	TGAGTGGAGG TCAA(C>t)GAGCA AGAAATTTCTT	762/763	233
A559T	11	95	rare	Substitute G>A at 91	GCAAGAATTT CTTTA(G>A)CAAG GTGAATAAC	762/763	233
R560T	11	95	0.40%	Substitute G>C at 95	ATTTCTTTAG CAA(G>C)GTGAAT AACTAA	762/763	233
R560K	11	95	rare	Substitute G>A at 95	GAATTTCTTT AGCAA(G>A)GTGA ATAACTAA	762/763	233
1898 + 1G>A	12	95	0.90%	Sub G>A after last Ex12	GAAATATTTG AAAG(G>A)TATGT TCCTTTGAAT	931/932	299
D648V	13	724	Nst Am (63%)	Substitute A>T at 177	AACTCATGGG ATGTG(A>T)TTCT TTCGACCAAT	955/884	360
2184 del A	13	724	0.70%	Delete A at 286	GACGAAACAA AAAAAaCAATC TTTTAAACAG AC	955/884	360
2184 ins A	13	724	rare	Insert A after 286	GACAGAAACA AAAAAaCAA TCCTTTAAAG CGAC	955/884	360
2789+5G>A	14b	38	1.10%	Sub G>A 5 one after last	CTCCTTGAA AGTGA(G>A)TATT CCATGTCCTA	885/886	374
3272-26A>G	17a	228	rare	Sub A>G 26 before 17b	TTTATGTTAT TTGCA(A>G)TGTT TTCTATGGAA A	782/901	414

Mutation	Exon	Exon size	Pop Freq	Location	Sequence Around Mutation Site (SEQ. ID. Nos. 128-191)	PRIMERS	Amp Sz.
3272-93T>C	17a	228	rare	Sub T>C 93 before 17b	ATTTGTGATA TGATTA(T>C)TCT AATTTAGTCT TT	782/901	414
R1066C	17b	228	rare	Substitute C>T at 57	AGGACTATGG ACAC TT(C>T)GTG CCTTCGGACG GC	782/901	414
L1077P	17b	228	rare	Substitute T>C at 91	TTACTTTGAA ACTC(T>C)GTTCC ACAAAGCTC	782/901	414
Y1092X	17b	228	0.50%	Substitute C>A at 137	CCAAC TGGTT CTTGTA(C>A)CTG TCAACACTGC G	782/901	414
M1101K	17b	228	Mut (65%)	Substitute T>A at 163	TGCGCTGGTT CCAAA(T>A)GAGA ATAGAAATGA T	782/901	414
R1152X	19	249	0.90%	Substitute C>T at 16	ATGCGATCTG TGAGC(C>T)GAGT CTTTAAGTTC	784/785	356
3659 del C	19	249	0.80%	Delete C at 59	AAGGTAAACC TACCAAGTCA ACCAAAACCAT ACA	784/785	356
3849 + 4 A>G	19	249	1.00%	Sub A>G 4 after last base	TCCTGGCCAG AGG GTG(A>G)GAT TTGAACACT	784/785	356
3849 10kb	19	10kb	1.40%	Sub C>T EcoR1 Fragment	ATAAAATGG(C>T) GAGTAAGACA	792/791	450
W1282R	20	156	rare	Substitute T>C at 127	AATAACTTTG CAACAG(T>C)GGA GGAAAGCCTT T	764/786	351
W1282X	20	156	2.10%	Substitute G>A at 129	AATAACTTTG CAACAGTG(G>A)A GGAAAGCCTT T	764/786	351
3905 ins T	20	156	2.10%	Insert T at 58	CTTTGTTATC AGCTTTT TTTG AGACTACTGA ACAC	764/786	351
4005 + 1 G>A	20	156	Manchester	Sub G>A after Exon 20	AGTGATACCA CAG(G>A)TGAGCA AAAGGACTT	764/786	351
N1303K	21	90	1.80%	Substitute C>G at 36	CATTTAGAAA AAA(C>G)TTGGAT CCCTATGAAC	756/793	396
N1303H	21	90	rare	Substitute A>C at 34	CATTTAGAAA A(A>C)ACTTGGAT CCCTATGAAC		